

### AGL Variant Allele Frequencies

8/28/2008

**Notes:** These summary statistics were compiled from clinical tests.

Only variants that are within the coding regions and about 50 base pairs of flanking sequence on each side are reported.

At this time, these summary statistics do NOT include insertion/deletion variants.

Exon	Left Flanking	Sequence Variation	Right Flanking	Coding Change	SNP Reference	Allele Frequencies
2	GATTTCAAATCCTCT	c.-10 A>G	GAAGCCAAAATGGGA	Pre coding	rs2307130	A-65.4% G-34.6%
3	GTAATTTAAGTCCTA	c.83-33 C>T	GATGAGTTTATTAAC	Intronic	rs2307129	C-74.8% T-25.2%
3	TGTAAACTTAATCTG	c.256 C>T	AACAATCTGGTTCAT	p.Gln86Stop		C-97.6% T-2.4%
3	ATTTCTTCAAGGGT	c.293+3 A>G	AGTCAGGTGTTTTGT	Intronic		A-99.5% G-0.5%
3	TTCCGATTAGGCCCA	c.112 A>G	CTTTACAGGGAAAAG	p.Thr38Ala		A-99.5% G-0.5%
3	TCTGGATTGGGAAAA	c.207 T>C	CCAACAGAAAGAGAA	p.Asn69Asn	rs2230305	T-97.6% C-2.4%
4	GAAAAGTTTTTGTTT	c.294-15 T>A	GTTTTTCCCTTAGA	Intronic		T-99.5% A-0.5%
4	ATAGTTGTGGACCCC	c.334 A>G	TTTTACGTGTTGGTG	p.Ile112Val		A-99.5% G-0.5%
5	TCTACAATCATACTG	c.664+1 G>A	TATGAGCTTCATTGA	Intronic		G-97.1% A-2.9%
5	CTAGGTCATGCTACT	c.521 C>T	CCTTGCCAATCAGTT	p.Ser174Phe		C-99% T-1%
5	TGGAATGATGTTGGA	c.595 C>T	AGCTAGTGGAAAAAT	p.Gln199Stop		C-99.5% T-0.5%
6	ATAGTAAATGGATCC	c.686 A>G	GGAACATCCAGAATG	p.Gln229Arg	rs17121403	A-99% G-1%
6	CACTTAAAACCTGCC	c.742 T>C	GGGTCTTAGACAGAG	p.Trp248Arg		T-99.5% C-0.5%
7	TCCAAAGCTTAAACT	c.894 C>T	TGGGAATTTTTCCAA	p.Leu298Leu	rs2230306	C-69.1% T-30.9%
8	TGCATTTAAGGTATC	c.959-18 G>A	TCTTTTCTTTCTTTT	Intronic	rs634880	A-63.5% G-36.5%
8	GATCCTGAATACAGA	c.1027 C>T	GGTTTGGCTGTACTG	p.Arg343Trp		C-98.6% T-1.4%
8	ACGACTTTCATACCA	c.1078 C>T	ATGAGTATGTAATGT	p.His360Tyr		C-99% T-1%
9	ATTAAATTCAGAGAA	c.1155 G>T	CATCGACTCATTAAC	p.Lys385Asn	rs28730701	G-99% T-1%
9	ATTCAGAGAAGCATC	c.1160 G>A	ACTCATTAACTATCA	p.Arg387Gln	rs17121464	G-99% A-1%
12	---TAATTGTTTTT	c.1424-44 A>G	TTTTATTTCTTGAAC	Intronic	rs2291638	A-76.4% G-23.6%
12	AGGTTCAGAAGTTTA	c.1437 C>A	CTAAGGAGAGAACTT	p.Tyr479Stop		C-99.5% A-0.5%
12	ACAGTGTTAAATTAC	c.1481 G>A	CTATGGGAATAAACC	p.Arg494His		G-99.5% A-0.5%
12	GTGTAAATTACGCT	c.1484 A>G	TGGGAATAAACCAGA	p.Tyr495Cys		A-99.5% G-0.5%
12	ATTTCCAGGGAGTAC	c.1571 G>A	TCTTGRTAACTGCCA	p.Arg524His		G-99.5% A-0.5%

12	AGGGAGTACRTCTTG	c.1577 A>G	TAAGTGCCTTGTGTT	p.Asp526Gly		A-99.5% G-0.5%
13	CTTTCTGTTACATTT	c.1612-32 A>G	TTTGTTACATTTGTC	Intronic		A-99.5% G-0.5%
13	TACATTTATTTGTTA	c.1612-24 C>A	ATTTGTCACTGTGCT	Intronic		C-97.6% A-2.4%
14	GAGTGTCCATTGTG	c.1899+1 G>A	TAAGCACCTAATCTT	Intronic		G-98.6% A-1.4%
15	CTCATCAGGTTTGT	c.2001+8 T>C	ATATGTTGTTTCTTA	Intronic	rs3736296	T-78.9% C-21.1%
16	GTGGTTTCTGAAGAA	c.2023 C>T	GGTTTTACACTAAGT	p.Arg675Trp		C-99.5% T-0.5%
16	TGGTTTCTGAAGAAC	c.2024 G>C	GTTTTACACTAAGTG	p.Arg675Pro		G-99.5% C-0.5%
20	GCTGTTGGAATTCTT	c.2590 C>T	GAAATCATCTGACAC	p.Arg864Stop		C-97.6% T-2.4%
20	AATTCCTTTTGCTTC	c.2681+1 G>T	TAAGTATGCCTTGT	Intronic		G-99.5% T-0.5%
21	TCAAGGTAAGCAAAT	c.2812+11 G>A	GAAGGATAGCTGAGC	Intronic	rs555929	A-58.7% G-41.3%
21	AGCCCTTAAATATGC	c.2802 A>C	GGTCTTCAAGGTAAG	p.Ala934Ala	rs34230588	A-99.5% C-0.5%
22	GTAATAATTTGAGAT	c.2885 C>G	TGGAGATTGGATGAT	p.Ser962Cys		C-99.5% G-0.5%
23	ATACCAAATTAACCTT	c.2950-21 T>A	CAAATTTATTTAAT	Intronic	rs2035961	T-57.2% A-42.8%
24	TAAAATATGTAAATC	c.3259+37 G>A	ATAGTATTCGCGGAA	Intronic	rs594249	A-57.2% G-42.8%
24	GCCCTAATGGATGTA	c.3199 C>T	CTTATAGGTTAAATG	p.Pro1067Ser	rs3753494	C-86.5% T-13.5%
24	TCTCTAGCTGCAGGT	c.3259+3 A>T	AGGAATTATGTACAA	Intronic		A-98.1% T-1.9%
24	TATGTACAAGGTTAA	c.3259+25 A>G	ATATGTAAATCRATA	Intronic		A-99.5% G-0.5%
24	ATGTACAAGGTTAAA	c.3259+26 A>G	TATGTAAATCRATAG	Intronic		A-99.5% G-0.5%
24	TACAAGGTTAAAATA	c.3259+29 T>C	GTAAATCAATAGTAT	Intronic		T-99.5% C-0.5%
24	TAAAATATGTAAATC	c.3259+38 G>A	ATAGTATTCGCGGAA	Intronic		G-98.1% A-1.9%
25	ATACTGCTGATTACT	c.3343 G>A	GACGCTATGTAGAAG	p.Gly1115Arg	rs7536086	G-93.3% A-6.7%
25	ATTAACATATTACTT	c.3260-20 T>C	GTTGTGTTTTTTTTG	Intronic		T-99% C-1%
25	TTAACATATTACTTT	c.3260-19 G>A	TTGTGTTTTTTTTGT	Intronic		G-97.6% A-2.4%
26	GTTTTTTTGT	c.3588+36 T>G	TTTTTTTT-----	Intronic	rs548255	T-97.1% G-2.9%
27	GAAGTCATACAGGAA	c.3619 G>A	CAATGCAAAAACACA	p.Ala1207Thr	rs11807956	G-98.1% A-1.9%
27	AAAATAGTACAAATT	c.3700+32 T>C	ATCAAGGTGATGAAA	Intronic	rs834575	T-96.2% C-3.8%
28	TATACCCTTCTTTAA	c.3836+45 G>A	AAAGCAC-----	Intronic	rs2274570	G-60.1% A-39.9%
28	CCCAGCCACACCAAG	c.3836+1 G>A	TAGTGTAATGTTAT	Intronic		G-99.5% A-0.5%
29	TAGAGATGGGTCTGC	c.3849 T>C	GTGGAAATTGTGGGC	p.Ala1283Ala	rs28730706	T-97.6% C-2.4%
30	TCTCATATGATGAGT	c.3980 G>A	GAACAGAAAAATACA	p.Trp1327Stop		G-95.7% A-4.3%
30	GAGCTTCAAGTCCTT	c.4115 G>A	GTGTGACTATCAGCT	p.Trp1372Stop		G-99.5% A-0.5%
31	AACTTTAGATCCAGA	c.4259+1 G>A	TAAGTTGGAATATAA	Intronic		G-99.5% A-0.5%
32	TTAACTTAAATTTCA	c.4260-12 A>G	TCATTTTGCAGTGAT	Intronic		A-97.4% G-2.6%
33	TTTCCCGACATTATG	c.4469 T>C	TCATCTTGAGAGGTA	p.Val1490Ala		T-99.5% C-0.5%
34	ACCAATGAGAATGCC	c.4525 C>T	AGTACTGTCCTTTCA	p.Gln1509Stop		C-99.5% T-0.5%
34	TACTGTCCTTTCAGC	c.4543 T>C	GTGAAACACAAGCCT	p.Cys1515Arg		T-99% C-1%